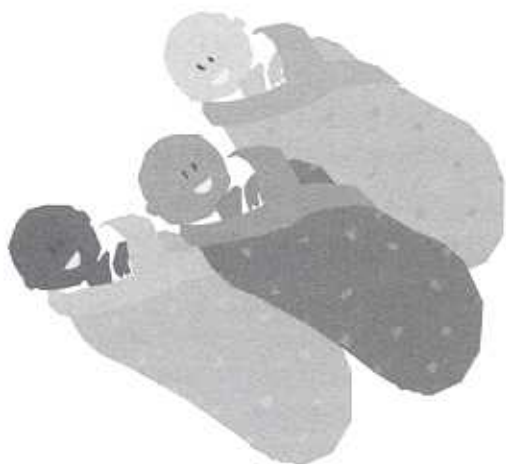




# **Your Baby and Newborn Screening**



**The Newborn  
Screening Program  
for the State of Indiana**



## WHAT IS THE NEWBORN SCREENING PROGRAM FOR THE STATE OF INDIANA?

Testing on blood for eight uncommon inherited disorders of body function. Also expanded testing for disorders detected using Tandem Mass Spectrometry (MS/MS).

## WHO IS SCREENED?

Your baby and every baby born in Indiana.

## WHEN IS IT DONE?

Just before you take your baby home from the hospital, or at 48 hours of age if born outside of a hospital.

## HOW IS IT DONE?

A tiny amount of blood from your baby's heel is sent to a special laboratory for testing.

## WHY IS IT DONE?

Because the screened disorders can be treated if detected early.

## WHAT ARE THE EIGHT ORIGINAL DISORDERS?

- Phenylketonuria (PKU)
- Galactosemia
- Hypothyroidism
- Homocystinuria (HCU)
- Maple Syrup Urine Disease (MSUD)
- Biotinidase Deficiency
- Congenital Adrenal Hyperplasia (CAH)
- Sickle Cell Disease/Hemoglobinopathies

## PHENYLKETONURIA (PKU)

Infants with PKU lack an enzyme which is needed by the body to break down phenylalanine, a component of protein in foods. A special diet low in phenylalanine can prevent the mental retardation and other effects of PKU.

The screening test for PKU is accurate only if your baby has taken adequate feedings containing protein, either formula or breast milk, before the test is obtained. For this reason, if your baby goes home before 48 hours of age, your hospital or physician will ask that a repeat specimen be collected within a few days.

## GALACTOSEMIA

Galactosemia is caused by a lack of an enzyme that prevents the body from processing galactose, a natural sugar found in milk (breast and cow's milk). Serious liver and brain damage, and even death may occur if the disorder is not detected early in life. A special milk-free diet can prevent or greatly reduce the effects of this disorder.

## HYPOTHYROIDISM

Hypothyroidism is caused by inadequate production of thyroid hormone that permits proper body and brain growth. Inadequate

thyroid hormone can result in mental retardation, but if detected early and hormone treatment started, normal growth and development take place.

## HOMOCYSTINURIA

Homocystinuria is caused by a problem in the way the body uses the amino acids, homocystine and methionine, which are present in proteins. The effects of the disorder may develop very slowly but are irreversible and can be life-threatening under certain circumstances. Early detection and treatment will reduce or prevent the complications.

## MAPLE SYRUP URINE DISEASE

The name of this disorder comes from the urine's distinctive maple syrup like odor and results from the body's inability to break down some components of protein in food (amino acids; leucine, isoleucine, and valine). A special diet must be started very early in life to prevent severe mental retardation and other physical problems.

## BIOTINIDASE DEFICIENCY

An absence of the enzyme biotinidase results in the body's inability to recycle biotin, a vitamin required for the proper functioning of several enzymes involved in glucose, amino acid, and fatty acid metabolism. Affected infants may develop seizures and problems with the brain's function. Early detection and treatment is beneficial to preventing the adverse effects of this disorder.

## CONGENITAL ADRENAL HYPERPLASIA

An absence of the enzyme 21-hydroxylase results in the body's inability to make normal amounts of the hormones, cortisol and aldosterone. These hormones are important in regulating the body's fluid and salt levels, and when they are not present the affected infant may become severely ill. This disorder may also result in the production of excess testosterone, the male hormone, which may cause an affected female infant's genitalia to look more like a male. Early detection and medical treatment can prevent severe illness or death.

## SICKLE CELL DISEASE AND HEMOGLOBINOPATHIES

Sickle Cell Disease is caused by a defect in the structure of hemoglobin, the oxygen-carrying component of blood. This defect causes the red blood cells to become rigid and sickle-shaped, resulting in problems with blood circulation, oxygen delivery to tissues, and anemia. Although there is no specific treatment for this disease, steps can be taken to prevent complications if the disorder is detected early in life.

## WHAT ARE THE DISORDERS DETECTED USING TANDEM MASS SPECTROMETRY (MS/MS)?

- Fatty Acid Oxidation Disorders
- Organic Acid Disorders
- Other Amino Acid Disorders

## Fatty Acid Oxidation Disorders

This group of disorders include conditions that interfere with the body's ability to turn fat into energy. The most common disorder of this group is Medium Chain Acy-CoA Dehydrogenase Deficiency (MCAD). Avoidance of Fasting is necessary with most of these disorders.

## Organic Acid Disorders

These disorders are characterized by the body's inability to break down amino acids and other metabolites normally used by the body. As a result, toxic products accumulate within the body causing organ damage and even death. Early detection can help prevent some adverse effects of these disorders.

## Other Amino Acid Disorders

These disorders include tyrosinemia and disorders of the Urea Cycle. As with the Organic Acid disorders, infants with these disorders lack the ability to break down certain metabolites. As a result, toxic products accumulate within the body causing organ damage and even death. Early detection can help reduce complications.



Expanded screening using Tandem Mass Spectrometry allows for multiple disorders to be screened for at the same time. However, some of these disorders may not be detected through newborn screening due to timing of the disease and individual genetic variability.

## How Is The Testing Done?

Just before you take your baby home from the hospital, the hospital will obtain a few drops of blood from your baby's heel. The blood is absorbed into a piece of special

paper, which is sent to the Newborn Screening Laboratory for testing to determine if your infant has any of the eight genetic disorders, or additional conditions evaluated with Tandem Mass Spectrometry (MS/MS).

It is important to note that the tests that are carried out are **SCREENING TESTS**, not diagnostic tests. The tests are designed to hopefully not miss a truly affected infant, but they are so sensitive that some non-affected infants will have falsely positive results. Actual diagnosis or the determination of whether one of the disorders is actually present must be made by additional tests which will be performed should any of the screening tests give positive results.

## What Are The Chances That My Baby Will Have One Of The Disorders?

These disorders are uncommon to rare in occurrence. Sickle Cell Disease occurs mainly in blacks, among whom the incidence is one case per 600 births. Hypothyroidism occurs at a rate of one case per 3000 births. The other disorders are less common and vary in occurrence from one case in 10-12,000 births (PKU and CAH); one case in 30-50,000 births (Galactosemia and Biotinidase Deficiency); one case in 100-250,000 births (Homocystinuria and MSUD), to one case in 20-30,000 (Fatty Acid Oxidation and Organic Acid).

## But my Baby Seems Very Healthy, Are The Tests Still Necessary?

YES! Most infants with these disorders show no obvious signs of the disease at birth. Most of the disorders result from problems with how the body produces or processes important chemical components inside the body, which usually do not result in any initial effect that can be seen. These "invisible" problems can only be detected using the special screening tests performed in the Newborn

Screening Laboratory. By testing every infant shortly after birth, we can be sure that each infant who has one of these disorders will be identified and started on treatment early before serious effects occur.

### **My Baby has One Of These Disorders, Can It be Cured?**

No, not really. It cannot be cured since these are genetic or inherited disorders; just as eye color or height can't be permanently changed. However, the serious effects of the disorders can be lessened – and often completely prevented – if a special diet or other medical treatment is started early.

**Early detection and treatment is very important!**

### **What Happens If One Of The Tests Results is "Positive"?**

If any of the tests show an abnormal result, the Newborn Screening Laboratory will contact your hospital and doctor immediately to request another blood sample. You will be asked to bring your baby in for a repeat test. Do so as soon as possible, as prompt action can be very important to your baby.

If you don't have a telephone, it will be helpful to leave the phone number of a neighbor or relative with the doctor. You can also help by notifying your doctor immediately if you move soon after your baby is born. Then if your child should need to be retested, your doctor will know where to reach you. Remember, time may be of great importance.

### **If A Retest Is Necessary, Does This Mean That My Baby Is Sick?**

No, not necessarily. Retesting may be requested for a number of reasons, many of which are related to when and how the newborn screen specimen was collected, i.e. too early, not enough blood collected, etc.

Also, the screening tests are very sensitive so truly affected infants will not be missed, and thus some non-affected infants will have falsely positive results. However, while taking your infant in for retesting can be inconvenient and worrisome, it is important that every infant receive the benefit of thorough screening for all eight disorders.

### **Will I Receive A Report Of The Test Results?**

Your doctor and hospital or clinic will be informed of the results with special emphasis placed on any "positive" or problem results. Generally, parents are notified only if there is a problem, but you should ask about the results when you take your baby to the doctor for a regular check-up. Remember, even if an infant is free of these disorders, there may be other medical problems which cannot be detected by this screening.



**It is very important for your baby to have regular check-ups and good general medical care.**

## QUESTIONS?

### Parents Should Contact:

Your private physician



### Maternal and Child Health Division

Genetic Disease or  
Newborn Screening Program  
Indiana State Department of Health  
2 North Meridian St., Suite 700  
Indianapolis, IN 46204  
317-233-1254

### Or Have Your Doctor Contact:

#### Newborn Screening Laboratory

Riley Hospital for Children  
702 Barnhill Drive, Rm 0940  
Indianapolis, IN 46202

### For Questions related to PKU, MSUD Galactosemia, Homocystinuria, Biotinidase Deficiency & Expanded Screening

#### Department of Pediatrics, Metabolism Section

Riley Hospital for Children  
702 Barnhill Drive, Rm A-36  
Indianapolis, IN 46202

### For Questions related to Hypothyroidism and Congenital Adrenal Hyperplasia

#### Department of Pediatrics, Endocrinology Section

Riley Hospital for Children  
702 Barnhill Drive, Rm A-584  
Indianapolis, IN 46202

### For Questions related to Sickle Cell Disease

#### Department of Pediatrics, Hematology Section

Riley Hospital for Children  
702 Barnhill Drive, Rm 2720  
Indianapolis, IN 46202